



## NBEAL2 gene

neurobeachin like 2

### Normal Function

The *NBEAL2* gene provides instructions for making a protein whose function is unclear. The protein appears to be critical for the normal development of platelets, which are blood cell fragments involved in blood clotting.

Platelets are produced in bone marrow, the spongy tissue in the center of long bones that produces most of the blood cells the body needs. Platelets are formed from large precursor cells known as megakaryocytes. Within these cells, the NBEAL2 protein is thought to play a role in the development of sacs called alpha-granules, which are the most abundant components of platelets. Alpha-granules contain growth factors and other proteins that are important for blood clotting and wound healing. In response to an injury that causes bleeding, the proteins stored in alpha-granules help platelets stick to one another to form a plug that seals off damaged blood vessels and prevents further blood loss.

### Health Conditions Related to Genetic Changes

#### gray platelet syndrome

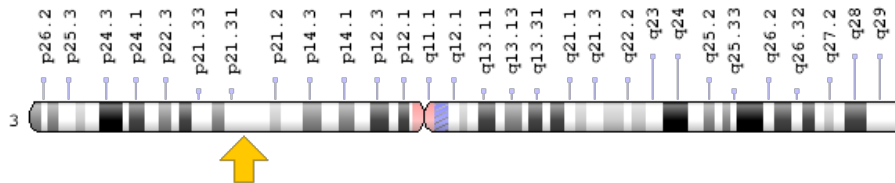
At least 35 mutations in the *NBEAL2* gene have been found to cause gray platelet syndrome, a disorder associated with abnormal bleeding. Most people with gray platelet syndrome also develop a condition called myelofibrosis, which is characterized by the buildup of scar tissue (fibrosis) in the bone marrow that prevents it from making enough normal blood cells.

Mutations in the *NBEAL2* gene disrupt the normal production of alpha-granules in megakaryocytes. Without alpha-granules, platelets are abnormally large and fewer in number than usual (macrothrombocytopenia). The abnormal platelets also appear gray when viewed under a microscope, which gives this condition its name. A lack of alpha-granules impairs the clumping of platelets in response to injury, increasing the risk of abnormal bleeding. Myelofibrosis is thought to occur because the growth factors and other proteins that are normally packaged into alpha-granules leak out into the bone marrow. The proteins lead to fibrosis that affects the bone marrow's ability to make new blood cells.

## Chromosomal Location

Cytogenetic Location: 3p21.31, which is the short (p) arm of chromosome 3 at position 21.31

Molecular Location: base pairs 46,979,683 to 47,009,704 on chromosome 3 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- BDPLT4
- GPS
- KIAA0540
- neurobeachin-like 2
- neurobeachin-like protein 2

## Additional Information & Resources

### Educational Resources

- University of Washington: Platelet Activation  
<http://courses.washington.edu/conj/bloodcells/platelets.htm>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28NBEAL2%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

### OMIM

- NEUROBEACHIN-LIKE 2  
<http://omim.org/entry/614169>

## Research Resources

- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=NBEAL2%5Bgene%5D>
- HGNC Gene Family: BEACH domain containing  
<http://www.genenames.org/cgi-bin/genefamilies/set/1230>
- HGNC Gene Family: WD repeat domain containing  
<http://www.genenames.org/cgi-bin/genefamilies/set/362>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=31928](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=31928)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/23218>
- UniProt  
<http://www.uniprot.org/uniprot/Q6ZNJ1>

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Reviewed: September 2014

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications  
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